Hypoplastic Left Heart Syndrome (HLHS)

Patient Information Series – What vou should know. what vou should ask.

What is a HLHS?

HLHS is a complex and rare congenital (present at birth) heart defect in which the left side of the heart is critically underdeveloped and cannot support blood flow through the heart to the body.

How does a HLHS happen?

The etiology of HLHS is unclear and considered to be multifactorial. It is a rare condition and happens in 0.1 to 0.25 per 1000 live births. Some babies have changes in their genes or chromosomes but in most cases, HLHS occurs by chance, with no clear reason. Less than one third of the babies with HLHS will have other problems in other organs.

How are chromosomes relevant to a HLHS?

Chromosomes are where most of our genetic information is kept. We usually have 46 of them matched in pairs: 23 come from one parent and the other 23 come from the other parent. For example, people with Down syndrome have an extra chromosome number 21. The common associated chromosomal abnormalities in these babies are Turner syndrome, trisomy 13 and trisomy 18. Except chromosomal abnormalities, multiple genetic syndromes have been reported, including Noonan's syndrome, Smith-Lemli-Opitz syndrome, Holt-Oram syndrome.

Should I have more tests done?

Many women will choose to have more tests done to know more about the condition of the baby. The tests available depend on where you are. Tests to ask about include:

- an **amniocentesis** to look for problems with the number of chromosomes and some of the problems within the chromosomes. This is done by removing small amount of amniotic fluid surrounding the fetus.
- a **genetic testing** to look for associated genetic syndromes.
- a **detailed fetal ultrasonography** to exclude associated problems in other organs.
- a **fetal echocardiography** - a specialized ultrasound of the heart of the baby during the pregnancy, which uses sound waves that “echo” off the structures of the fetus' heart.

What are the things to watch for during the pregnancy?

Babies with HLHS are at risk of some problems during the pregnancy. That is why most specialists will recommend regular ultrasound examination in every 4 to 6 weeks because impairment in heart function can progress over time or babies may be at risk of growth impairment during the pregnancy. In some babies with HLHS, in utero treatment may be possible and can, in select cases, promote growth of left side of heart or prevent high pressure and damage to the developing lung vasculature and improve the outcome.

What does it mean for my baby after it is born?

Babies with HLHS might not have trouble for the first few days of life while the normal openings in the heart (the patent ductus arteriosus and the patent foramen ovale) are not closed yet, but quickly develop signs after closure of these openings, such as bluish skin color, weak pulse and breathing problems. Pulse oximetry is a simple bedside test to determine the amount of oxygen in a baby's blood and low levels of oxygen can be a sign of HLHS before presenting any symptoms.

A doctor can find these signs or might hear a heart murmur (an abnormal sound resulted by abnormal flowing of blood). If these signs are present, the health care provider might request an echocardiogram or more tests to make a diagnosis. Echocardiography also is useful for following the child's health over time.

Babies who also have had the information within their chromosomes altered and HLHS may have even more problems after being born. This will depend on the nature of the genetic disorder.

There are treatments for some health problems related to HLHS such as medicines, nutrition and surgery. Soon after birth, performing multiple surgeries (in a particular order) are needed to increase blood flow to the organs and bypass the small left side of the heart. These surgeries do not cure HLHS but try to restore heart function and pump blood to baby's organs.

When they grow up, infants who have a HLHS and survive may have lifelong problems. They will need regular follow-up visits by a cardiologist. If the HLHS is very complex, or the heart becomes weak, a heart transplant may be indicated. Babies who receive a heart transplant will need medicines for the rest of their lives to prevent rejection of the new heart by their body.

In fact, many survivors face the wide range of neurodevelopmental challenges. Children who undergo cardiac surgery in the first year after birth are at higher risk for developmental, learning and behavioral problems later in their life. Multiple factors can influence neurodevelopment in these children, including associated neurological anomalies, genetic conditions and surgical techniques.

Will it happen again?

If no other genetic reason is found to explain the HLHS, the risk of this happening again is very low. If there is a genetic reason, this will determine the risk, and a consultation with a genetic specialist may be helpful to help sort this out.

If in the family there is one child with HLHS, the risk of having another baby with a similar condition is higher (the risk of recurrence in future pregnancies, estimated to be 2-4%) but in families with two affected children the risk increases to 25%.

What other questions should I ask?

- Does this look like a severe HLHS?
- How often will I have ultrasound examinations done?
- Is surgery during the pregnancy available?
- Is my baby one that could benefit from surgery inside the womb?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?

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